A 13-year-old boy with very high LDL-C (679 mg/dL) presented with planar xanthomas.

He was found to have a β-sitosterol level of 53.5 mg/L (normal < 3.3 mg/L).

He was a compound heterozygote for two ABCG8 mutations (p.N409D and an intron 11+2 T>A splice site mutation).

On a low cholesterol and plant-sterol diet, his LDL-C decreased to 485 mg/dL (-29%) and β-sitosterol to 44.6 mg/L (-27%).

On atorvastatin 20 mg/day, his LDL-C decreased to 299 mg/dL (-38%).

With added ezetimibe 10 mg/day, his LDL-C normalized to 60 mg/dL (-80% further decrease); and his β-sitosterol decreased to 14.1 mg/L (-68% further decrease).